Unit 5
Heredity

Chapter 11: Mendel and the Gene Idea

Mendel

- When Mendel crossed contrasting, true-breeding white- and purple-flowered pea plants
  - All of the F_1 hybrids were purple
  - No “blending”
- When Mendel crossed the F_1 hybrids
  - Many of the F_2 plants had purple flowers, but some had white
  - Mendel discovered a ratio of about 3:1 purple to white flowers in the F_2 generation

- Mendel reasoned that the heritable factor (**gene**) for white flowers was hidden or masked in the presence of the purple-flower factor
- He called
  - the purple flower color **dominant**
  - the white flower color **recessive**
- The factor for white flowers was not diluted or destroyed because it reappeared in the F_2 generation

Mendel determined

1. Alternative versions of genes account for variations in inherited characters
   - Now called **alleles**
2. For each character, an organism inherits two alleles, one from each parent
3. If the two alleles at a locus differ, then the **dominant allele** determines the organism's appearance, and the **recessive allele** has no noticeable effect on appearance
4. The two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes
   - Now known as the **law of segregation**

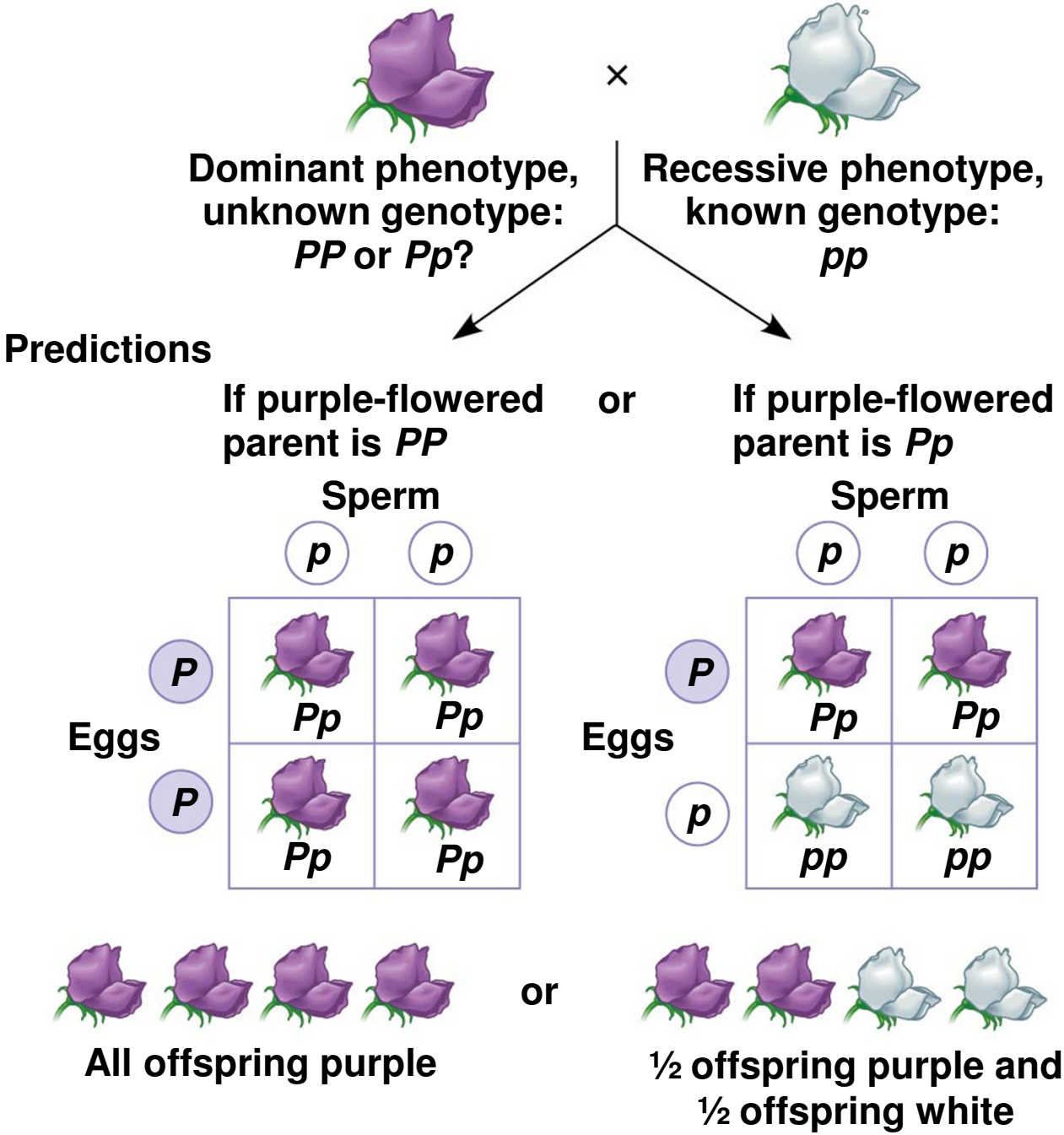
Useful Genetic Vocabulary

- **Homozygous** =
  - An organism with two identical alleles for a gene
  - True-breeding
- **Heterozygous** =
  - An organism that has two different alleles for a gene
  - NOT true-breeding
- **Phenotype** =
  - Physical appearance
- **Genotype** =
  - Genetic makeup

Crosses

- How can we tell the genotype of an individual with the dominant phenotype?
  - Such an individual could be either homozygous dominant or heterozygous
- The answer is to carry out a **testcross**
  - Breeding the mystery individual with a homozygous recessive individual
- If any offspring display the recessive phenotype, the mystery parent must be heterozygous

Figure 11.7


Technique
×
Dominant phenotype, unknown genotype: PP or Pp?
Recessive phenotype, known genotype: pp
Predictions
If purple-flowered parent is PP
or
If purple-flowered parent is Pp
Sperm
p
p
P
P
Eggs
Pp
Pp
Pp
Pp
Sperm
p
p
P
p
Eggs
Pp
Pp
pp
pp
Results
or
All offspring purple
½ offspring purple and ½ offspring white

- **Monohybrid cross** =
  - A cross between 2 organisms that are both heterozygous for 1 trait
  - Results in a phenotypic ratio of 3:1
- **Dihybrid cross** =
  - A cross between 2 organisms that are both heterozygous for 2 traits
  - Results in phenotypic ratio of 9:3:3:1
  - Can determine whether two genes are linked

The Law of Independent Assortment

- The results of Mendel's dihybrid experiments are the basis for the **law of independent assortment**
  - It states that each pair of alleles segregates independently of each other pair of alleles during gamete formation
- This law applies to genes on different, nonhomologous chromosomes or those far apart on the same chromosome
  - Genes located near each other on the same chromosome tend to be inherited together or "linked"

Practice Problems

- How many unique gametes could be produced through independent assortment by an individual with the following genotype?
  - AA = 1
  - Aa = 2
  - AaBb = 4
  - AaBbCcDdEe = 32
    - 2 x 2 x 2 x 2 x 2
  - AABbccDDee = 2
    - 1 x 2 x 1 x 1 x 1
    - ABcDe OR AbcDe

- AabbCcddEe = 8
- 2 x 1 x 2 x 1 x 2

~AbCdE ~abCdE

~AbcdE ~abcdE

~Abcde ~abcde

~AbCde ~abCde

Laws of Probability

- Ex: Dihybrid cross (YyRr x YyRr), what is the probability offspring are:
  - YYRR
    - ¼ (Probability of YY) **x** ¼ (RR) = 1/16
  - YyRR
    - ½ (Probability of Yy = ¼ **+** ¼) **x** ¼ (RR) = 1/8
  - YyRr
    - ½ **x** ½ = 1/4

- Ex: What is the probability that following pairs of parents will produce the indicated offspring?
  - AABBCC x aabbcc → AaBbCc
    - 1 x 1 x 1 = 1
  - AABbCc x AaBbCc → AAbbCC
    - ½ x ¼ x ¼ = 1/32
  - AaBbCc x AaBbCc → AaBbCc
    - ½ x ½ x ½ = 1/8
  - aaBbCC x AABbcc → AaBbCc
    - 1 x ½ x 1 = ½

Other Patterns of Inheritance

- **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are identical
- In **incomplete dominance**, the phenotype of heterozygote is an intermediate somewhere between the phenotypes of the two parental varieties
  - Ex: Flower color in snapdragons
    - RR = Red, WW = White, RW = Pink
- In **codominance**, the phenotypes of both alleles are fully expressed in the heterozygote
  - Ex: Coat color in cattle
    - RR = Red, WW = White, RW = Roan (Red AND White)

- **Multiple Alleles**
  - A gene that has more than 2 *alleles*
  - EX: the four phenotypes of the ABO blood group in humans are determined by three alleles of the gene: I^A, I^B, and I

    I^AI^A or I^Ai = Type A

    I^BI^B or I^Bi = Type B

    I^AI^B = Type AB

    ii = Type O
- **Polygenic inheritance**
  - Trait controlled by 2 or more *genes*
    - An additive effect of two or more genes on a single phenotype resulting in a continuum
  - Ex: Skin color in humans

Pedigree Analysis

- A **pedigree** is a family tree that describes the interrelationships of parents and children across generations
- Inheritance patterns of particular traits can be traced and described using pedigrees
- Pedigrees can also be used to make predictions about future offspring
- Key
  - Square = male; Circle = female
  - Shaded = affected
  - Half shaded = carrier

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CHAPTER 12: THE CHROMOSOMAL BASIS OF INHERITANCE

Sex-linked genes

- In humans, females are XX, and males are XY
- A gene that is located on either sex chromosome is called a **sex-linked gene**
  - **Y-linked genes**
    - There are few of these
    - Passed from father to all sons
  - **X-linked genes**
    - Fathers pass X-linked alleles to ALL of their daughters but NONE of their sons!
    - Mothers can pass X-linked alleles to both sons and daughters

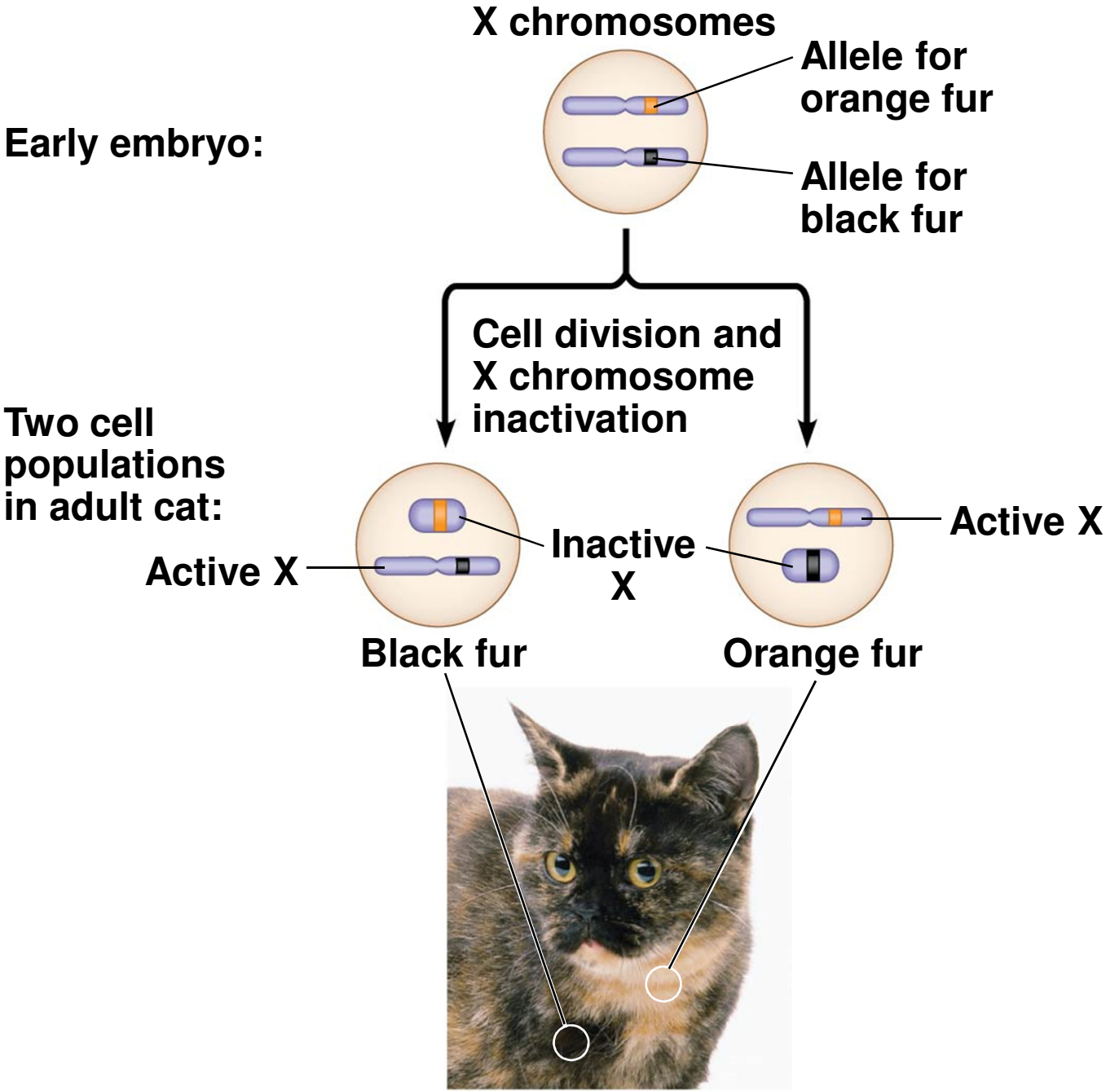
- For a recessive X-linked trait to be expressed
  - A female needs two copies of the allele (homozygous)
  - A male needs only one copy of the allele (***hemizygous***)
- X-linked recessive disorders are much more common in males than in females
  - Any male receiving the recessive allele from his mother will express the trait
  - But females can inherit the trait from an affected father and a mother who is a carrier

- Some disorders caused by recessive alleles on the X chromosome in humans
  - Color blindness (mostly X-linked)
  - **Duchenne muscular dystrophy**
    - Progressive weakening and loss of muscle tissue
  - **Hemophilia**
    - Absence of blood clotting proteins resulting in excessive bleeding following injury

X Inactivation in Female Mammals

- In mammalian females, one of the two X chromosomes in each cell is randomly inactivated during embryonic development
- The inactive X condenses into a **Barr body**
- If a female is heterozygous for a particular gene located on the X chromosome, she will be a mosaic for that character

Figure 12.8


X chromosomes
Allele for orange fur
Allele for black fur
Early embryo:
Cell division and X chromosome inactivation
Two cell populations in adult cat:
Active X
Inactive X
Active X
Black fur
Orange fur

Linked genes

- Genes that tend to be inherited together because they are located near each other on the same chromosome are called **linked genes**
- Ex: Morgan found that body color and wing size are usually inherited together in specific combinations
  - He reasoned that since these genes did not assort independently, they were on the same chromosome
- Can use Chi-Square Test to determine if genes are linked
  - Null hypothesis: Genes assort independently (predict resulting phenotypic ratio)
  - If null is rejected, then the genes may be linked

- Morgan discovered that even when two genes were on the same chromosome, some recombinant phenotypes were observed
- He proposed that some process must occasionally break the physical connection between genes on the same chromosome
- That mechanism was the **crossing over** between homologous chromosomes
  - End portions of two nonsister chromatids trade places during Prophase I of Meiosis
  - Accounts for recombination of linked genes

Linkage maps

- The recombination frequency depends on the distance between genes on a chromosome
  - The farther apart two genes are, the higher the probability that a crossover will occur between them and therefore the higher the recombination frequency
- A **linkage map** is a genetic map of a chromosome based on recombination frequencies
- Distances between genes can be expressed as **map units**
  - One map unit represents a 1% recombination frequency

- Genes that are far apart on the same chromosome can have a recombination frequency near 50%
  - Such genes are physically linked by being on the same chromosome
  - But genetically unlinked and sort independently
  - Thus behave as if found on different chromosomes!

Abnormal Chromosome Number

- In **nondisjunction**, pairs of homologous chromosomes do not separate normally
  - Occurs during anaphase of meiosis
  - As a result, one gamete receives two of the same type of chromosome, and another gamete receives no copy
- **Monosomy** = Zygote has only one copy of a particular chromosome
- **Trisomy** = Zygote has three copies of a particular chromosome
  - Ex: Down syndrome (Trisomy 21)

Aneuploidy of Sex Chromosomes

- Nondisjunction of sex chromosomes produces a variety of aneuploid conditions
  - Tend to be less severe than autosomal aneuploids
- Klinefelter syndrome is the result of an extra chromosome in a male, producing XXY individuals
- Females with trisomy X (XXX) have no unusual physical features except being slightly taller than average
- Monosomy X, called Turner syndrome, produces X0 females, who are sterile
  - It is the only known viable monosomy in humans!

- **Polyploidy** is a condition in which an organism has more than two complete sets of chromosomes
  - Triploidy (3*n*) is three sets of chromosomes
  - Tetraploidy (4*n*) is four sets of chromosomes
- Polyploidy is common in plants, but not animals

Alterations of Chromosome Structure

- Errors in meiosis or damaging agents like radiation can cause breakage of a chromosome
- This can lead to four types of changes in chromosome structure
  - **Deletion** removes a chromosomal segment
    - Missing certain genes
  - **Duplication** repeats a segment
  - **Inversion** reverses orientation of a segment within a chromosome
  - **Translocation** moves a segment from one chromosome to another

Figure 12.14a

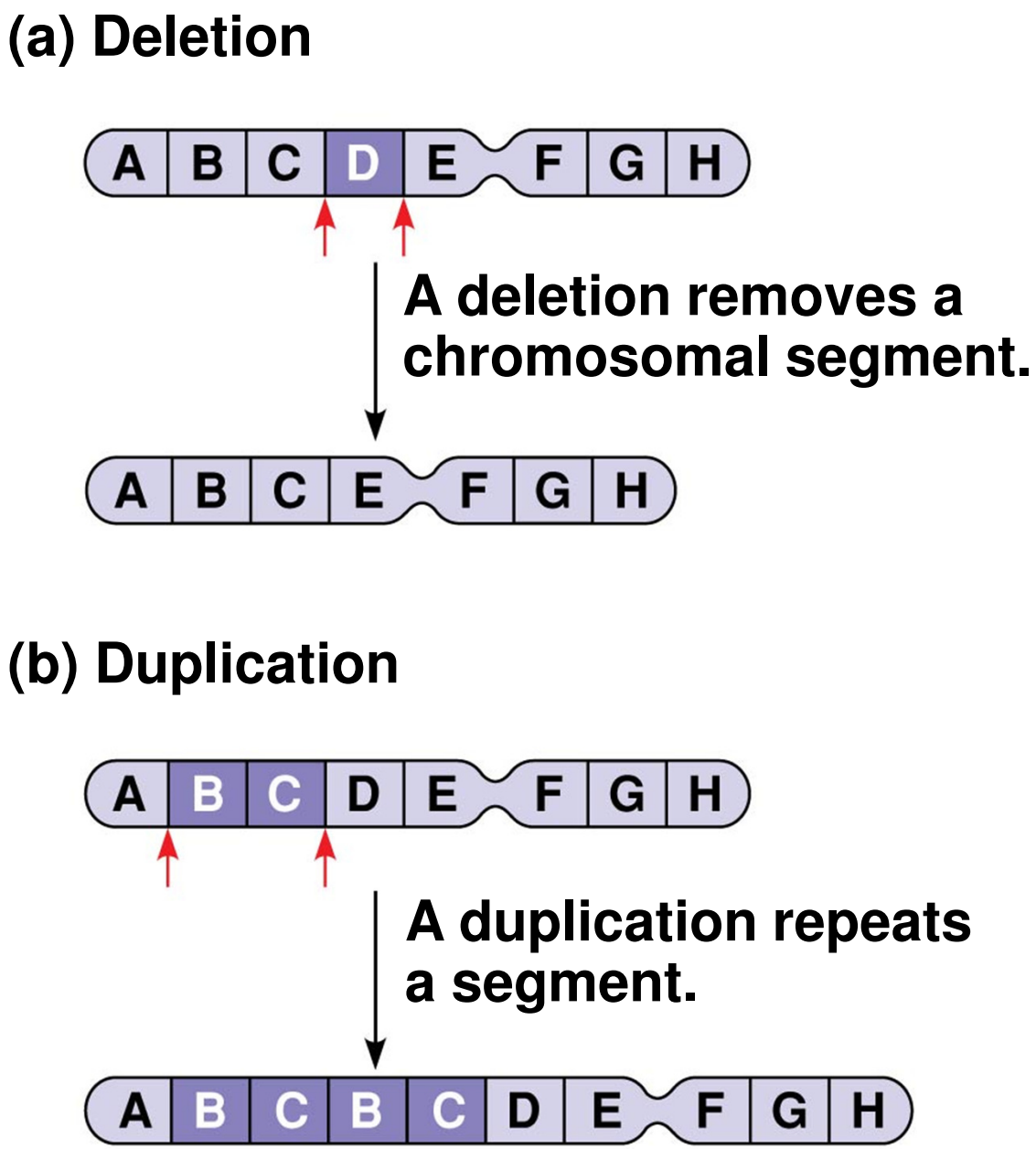


Figure 12.14b